

## Supplementary material

### **SPATA7: Evolving Phenotype from Cone-Rod Dystrophy to Retinitis Pigmentosa**

Matsui, McGuigan, Gruzensky, Aleman, Schwartz, Sumaroka, Koenekoop, Cideciyan & Jacobson

**Supplementary Table 1.** Retinal disease genes sequenced using Illumina HiSeq 2000 (Wang et al., 2013)

ABCA4	CHM	IDH3B	PAX2	RP2
ABCC6	CLN3	IMPDH1	PCDH15	RP9
ADAM9	CLRN1	INPP5E	PDE6A	RPE65
AHI1	CNGA1	INVS	PDE6B	RPGR
AIPL1	CNGA3	IQCB1	PDE6C	RPGRIP1
ALMS1	CNGB1	JAG1	PDZD7	RPGRIP1L
ARL6	CNGB3	KCNJ13	PEX1	RS1
ARMS2	CNNM4	KCNV2	PEX2	SAG
ATXN7	COL11A1	KLHL7	PEX7	SEMA4A
BBS1	COL2A1	LCA5	PGK1	SNRNP200
BBS10	COL9A1	LRAT	PHYH	SPATA7
BBS12	CRB1	LRP5	PITPNM3	TEAD1
BBS2	CRX	MERTK	PRCD	TIMM8A
BBS4	CYP4V2	MFRP:C1QTNF5	PROM1	TIMP3
BBS5	DFNB31	MKKS	PRPF3	TLR3
BBS7	DMD	MTTP	PRPF31	TLR4
BBS9	EFEMP1	MYO7A	PRPF8	TMEM126A
BEST1	ELOVL4	NDP	PRPH2	TOPORS
C2	ERCC6	NPHP1	RAX2	TREX1
C2ORF71	EYS	NPHP3	RB1	TRIM32
C3	FBLN5	NPHP4	RBP3	TRPM1
CA4	FSCN2	NR2E3	RBP4	TSPAN12
CABP4	FZD4	NRL	RD3	TTC8
CACNA1F	GNAT1	NYX	RDH12	TTPA
CACNA2D4	GNAT2	OAT	RDH5	TULP1
CC2D2A	GPR98	OFD1	RGR	UNC119
CDH23	GRK1	OPA1	RGS9	USH1C
CDH3	GRM6	OPA3	RGS9BP	USH1G
CDHR1	GUCA1A	OPN1LW	RHO	USH2A
CEP290	GUCA1B	OPN1MW	RIMS1	VCAN
CERKL	GUCY2D	OPN1SW	RLBP1	WFS1
CFB	HMCN1	OTX2	ROM1	
CFH	HTRA1	PANK2	RP1	

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**Supplementary Figure 1.** Published, disease-associated *SPATA7* gene exonic (below the diagram) and intronic (above the diagram) mutations on a scheme of the genomic structure. Numbers indicate exons. Black box indicates the mutation identified in the reported patient.

